Fall 2008

Newborn Screening 411

RCHSD
Newborn Screening
Area Service Center

Events

Our 1st Anniversary! In July we celebrated our one year anniversary with Rady Children's Hospital. As many of you know, the Area Service Center moved from the University of California, San Diego, to Rady Children's Hospital in July 2007. With UCSD and Rady Children's pediatric amalgamation program, this "best fit" means we still get to enjoy a close relationship with the specialists and folks at UCSD, but are housed within a CCS-approved multidisciplinary center.

Maternal PKU Camp! Our staff took advantage of an opportunity to meet a few of the very patients that the California Newborn Screening Program has helped. We spent the first week in August assisting at the 13th Maternal PKU Camp at Cal State Northridge; this week-long summer activity for women with PKU is hosted by California Department of Public Health and offers learning experiences and classes that teach women with PKU how to care for themselves by planning carefully for pregnancy leading to a healthy outcome for their infant. Women with PKU must be extra careful to control the amount of phenylalanine they consume during pregnancy since high levels can have negative effects on their fetus. More information can be obtained by visiting the California Department of Public Health website http://www.cdph.ca.gov.

Newborn Screening Month

On September 11, 2008 the Area Service Center hosted an Open House at Rady Children's Hospital. We enjoyed a great turn-out with some of you coming from as far away as Los Angeles and Riverside County. Without all of you, this important program could not meet its essential mission of early detection, diagnosis, and treatment of babies with screened-for disorders. We would like to recognize Charlotte for organizing this very successful event and look forward to seeing all of you again next year.

From left to right: Kimberly, Gail, Barbara, Stevie, Charlotte, Kristin and George.

RCHSD

Newborn Screening

Area Service Center

LOCATED AT:
7920 Frost Street, Suite 302
San Diego, CA 92123

MAILING ADDRESS:
3020 Children's Way
Mail Code 5116
San Diego, CA 92123

PHONE:
(858) 966-8708

FAX:
(**858) 966-8709**

For more information visit:
www.dhs.ca.gov/NBS/

"A baby is born with a need to be loved and never outgrows it."
-Frank A Clark

Sickle Cell Awareness Month

September is also Sickle Cell Awareness Month. Sickle cell disease is a hereditary disorder that affects the red blood cells. The defects in the hemoglobin cause the red blood cells to become hard and sticky, and change to a banana (sickle) shape. These sticky, sickled cells can clog small blood vessels, which does not allow blood to bring oxygen to the tissues, causing pain and damage.

Affected infants are susceptible to overwhelming bacterial infections that, if not promptly treated, can cause death. To prevent serious infection, prophylactic penicillin and special vaccines are given. Additionally, their parents are provided information and instruction about preventive health measures and how to recognize early warning signs of illness so that they can seek early medical attention. New treatments and good preventive therapies have improved the life expectancy and quality of life for people with sickle cell disease.

ARUP Guidelines for Galactosemia

Starting in August, the Associated Regional and University Pathologists, Inc. Laboratory (ARUP) at the University of Utah entered into an agreement with the California Genetic Disease Screening Program to perform galactosemia confirmatory testing. New guidelines and procedures for specimen collection, transport, and analysis have been sent to metabolic specialists and collection facilities. The Area Service Center will continue to coordinate collection and shipping of specimens by phone and by faxed instructions to primary care providers and collection facilities. The most significant change is the addition of DNA analysis, which will aid the specialist in making a diagnosis.

ARUP will measure Galactose-1-phosphate uridyl transferase (GALT). GALT levels below the designated cut-off will then have genotyping (i.e., DNA testing) to identify nine of the most common galactosemia gene mutations. The results mailer sent to the baby's primary care provider as well as the collecting facility will, as always, cite the test results and an interpretation. Newborns with confirmatory results consistent with classical galactosemia or Duarte galactosemia will be referred to a Metabolic Center for confirmation of diagnosis and treatment when indicated.

Laboratories asked to collect confirmatory specimens will receive a "Manual Order" requisition form which will be partially filled out by Area Service Center Staff and should be completed by the laboratory staff. That *manual order* will be accompanied by an instruction sheet with complete details on packaging and shipping. To avoid payment delays, **do not** use ARUP computer ordering.

If you have any questions about these changes or in general regarding Newborn Screening, please do not hesitate to call us at 1-800-793-1313. Instructions and technical bulletins from ARUP are available at http://www.chsd.org/newbornscreening.

TRF Update

In the section of the TRF labeled "Nursery Type," please mark either "NICU" or "Regular Nursery" if the baby is still in the hospital when the specimen is collected. Specimens collected from babies in the Mother-Baby Unit or in Couplet Care should be marked "Regular Nursery." "Other " should be marked for out-of-hospital births and for babies who are outpatients (e.g., discharged babies who must have another newborn screen).